



FOP diagnostic case series

When should we think FOP?

Delayed diagnosis and misdiagnosis are common in FOP, so establishing a prompt, correct diagnosis is key for avoiding inadvertent harm.

Click the buttons below to explore different case studies or review the key information about FOP



Case 1

FIND OUT MORE



Case 2

FIND OUT MORE



Case 3

FIND OUT MORE



Key information

FIND OUT MORE



A two-year-old was taken to the clinic as his parents were concerned that he was **not yet walking**. He had **no allergies** or **health conditions**, but his clinical notes did mention that at birth he had **unusually shaped toes**.



Initial consultation with pediatrician

A two-year-old was taken to the clinic as his parents were concerned that he was **not yet walking**. He had **no allergies** or **health conditions**, but his clinical notes did mention that at birth he had **unusually shaped toes**.

- Physical examination revealed that as well as not walking, the child was **'scooting'** instead of crawling
- The pediatrician also notices the child is **unable to turn his head** to one side
- Parents are advised that all children develop differently, but as a cautionary measure, would **refer to a specialist**



Who would you refer this patient to?

Click on the panels to discover why each of these specialists may be considered

NEUROLOGY

ORTHOPEDICS

RHEUMATOLOGY





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NEUROLOGY



The inability of a two-year-old to walk and turn his head may be indicative of neurological issues, which a neurologist would be well placed to investigate further.^{1,2}



Initial consultation with pediatrician

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ORTHOPEDICS



Orthopedics specialize in disorders affecting the **musculoskeletal system** — the **limited range of motion** in the neck and delayed ability to walk could be investigated further by an orthopedic specialist.¹



Initial consultation with pediatrician

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RHEUMATOLOGIST



Rheumatologists specialize in **autoimmune and inflammatory disorders** — often these can present as **joint/range of motion problems**.^{1–3}



Referral to specialist (pediatric neurologist)

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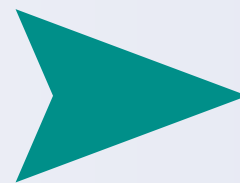
- In this case, the patient was **referred to a neurologist**
- Physical examination **excluded cervical dystonia**
- A **swelling** on the child's back was noticed, it was **hot to the touch** and caused pain
 - Parents reported this appearing shortly after the child **fell from a step**
- The clinician requested **analgesics and a follow-up appointment**



Referral to specialist (pediatric neurologist)

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- The clinician requested **analgesics and a follow-up appointment**



6 months later

- The child and his family did not attend the follow-up appointment due to a family bereavement
- The child then **developed a fever** and was taken to the **emergency department**
 - A **respiratory infection** with *pertussis* was suspected, and the patient was **prescribed antibiotics**
 - A **swelling on the shoulder** was also noted during examination
- Due to a **history of previous swellings**, the patient was **referred to rheumatology**



Referral to specialist (rheumatologist)

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Which tests might you recommend?

Click on the panels to discover why each of these specialists may be considered

**BIOCHEMICAL PROFILING
BLOOD TEST**

BIOPSY OF SWELLING

**BLOOD TEST FOR
INFLAMMATORY MARKERS**





Referral to specialist (rheumatologist)

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BIOCHEMICAL PROFILING



Different **biochemical markers** can be measured to identify the **functioning capacity** of several **organs** — for example, increased ALP can be associated with liver or bone health.¹



Referral to specialist (rheumatologist)

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BIOPSY OF SWELLING



Swellings can be initially suspected as **juvenile fibromatosis, lymphedema, or soft tissue sarcoma**,¹ in which case a biopsy would provide more information.*



Referral to specialist (rheumatologist)

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TEST FOR INFLAMMATORY MARKERS



The immune system can be activated by many triggers, from **injury** to **infection** to **auto-immune disorders**.¹

Measuring **inflammatory markers** can help **identify the cause** of an activated immune system.¹



Evaluation of inflammatory markers

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After visiting a pediatric neurologist, the patient **developed a fever** and was taken to the **emergency department**. As a result, he was given antibiotics to treat a **suspected infection**. A **swelling on the shoulder** was also noted, resulting in a **referral to a rheumatologist**.



Inflammatory marker results:

- Increased lymphocytes
- Increased IL- 3, IL-7 and GM-CSF

- The increase in inflammatory markers and history of limited joint mobility prompted the rheumatologist to seek the advice of a **multidisciplinary team**
- The team concluded that a **gene panel** for different **skeletal dysplasia conditions** should be carried out



Evaluation of gene panel (skeletal dysplasia)

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After visiting a pediatric neurologist, the patient **developed a fever** and was taken to the **emergency department**. As a result, he was given antibiotics to treat a **suspected infection**. A **swelling on the shoulder** was also noted, resulting in a **referral to a rheumatologist**. After consulting a multidisciplinary team, a **gene panel** was carried out, focusing on **skeletal dysplasia**.



Gene panel results:

- Mutation in the ALK2/ACVR1 gene
- c.617G>A



Diagnosis of FOP



Outcome

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Following a **confirmed diagnosis of FOP**, the patient's family was given a **list of recommendations** to follow (to avoid triggering flare-ups) and **genetic counselling**.

What could have been done differently?



Missed sign

The most important symptoms that lead to a diagnosis of FOP are **malformation of the great toes** (present from birth) and **soft tissue swellings**.¹



Missed sign

Stiffness of the neck is an early finding in most patients with FOP and may **precede heterotopic ossification**. This can prevent crawling and **babies may scoot** on their bottom instead.^{1,2}



Patient history

A five-year-old was taken to her primary care provider as she had **multiple swellings on her head**. Her parents stated that they had noticed **neck swellings** around two weeks ago, but these had **disappeared**. A physical examination revealed that the patient also had **shortened big toes**.

Click the buttons below to find out why

Referral to oncology

**Results from
oncology referral**





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Referral to oncology

Swellings could be **indicative of tumors**.¹

Oncologists would be best placed to identify whether the swelling is due to a tumor and which treatment would be most suitable.

Results from oncology referral



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Referral to oncology

Results from oncology referral

A **biopsy** and histological analysis of the swelling led the oncologist to diagnose the patient as having **aggressive juvenile fibromatosis**.

Surgical removal of the swellings was planned.



Preparation for surgery

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*Click to discover what
happened prior to
undergoing surgery*





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Pre-surgery MRI

- Revealed **calcification** within swellings
- Oncology worked with **orthopedic surgeon** for further consideration

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Orthopedic surgeon

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Click to discover the results of the genetic test





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Orthopedic surgeon

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Genetic testing

- Mutation within the *ACVR1/ALK2* gene
- **Diagnosis of FOP confirmed**



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Prior to the surgery, an **MRI scan** revealed that the **lesions were calcified**, prompting a referral to an **orthopedic specialist**. Genetic testing revealed a **mutation** in the **ACVR1/ALK2** gene, confirming the diagnosis of FOP.

What could have been done differently?



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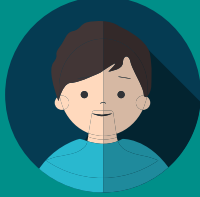
Harmful procedure

Biopsies should be avoided in people with FOP as the procedure can further exacerbate the disease.¹



Non-invasive investigation

Radiography and bone scans can be used to identify heterotopic ossification in patients with suspected FOP.²



Patient history



An eight-year-old boy presented at the clinic with a **severe mouth ulcer**, for which he was given an antibiotic mouthwash. During his follow-up appointment, his clinician noticed that the patient's **jaw movement was slightly restricted**.



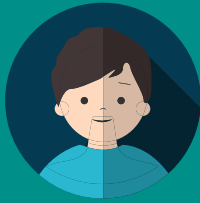
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DENTIST

PHYSIOTHERAPIST

MAXILLOFACIAL
SURGEON



Patient history



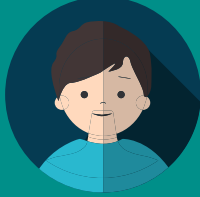
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DENTIST



Dentists can investigate whether jaw pain and restricted movement is due to actions such as grinding of the teeth.¹



Patient history



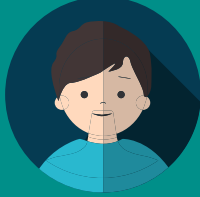
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PHYSIOTHERAPIST



Physiotherapists can massage the musculature around the joint and guide the patient through jaw-strengthening exercises.^{1*}



Patient history



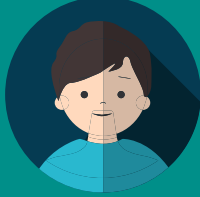
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MAXILLOFACIAL SURGEON



If the jaw is limited by congenital malformation of the joint, or other methods have failed to successfully treat the issue, a maxillofacial surgeon is specialized to treat the jaw with more invasive procedures.¹

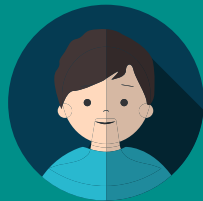


Patient history



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- In this case, the patient was **referred to a physiotherapist** to stretch the jaw and recommend jaw-strengthening exercises
- Six months later, the boy returned to the clinic with **stiffness and pain** in the back
- The clinician was told that the boy had recently **fallen over** after playing with some friends in the park
- Upon examination, **small swellings** in and around the **neck and spine** were observed



Patient history

An eight-year-old boy presented at the clinic with a **severe mouth ulcer**, for which he was given an antibiotic mouthwash. During his follow-up appointment, his clinician noticed that the patient's **jaw movement was slightly restricted**. He referred the patient to a **physiotherapist** to **stretch the jaw** and recommend exercises to **strengthen the jaw muscles**.

Six months later, the patient returned to the clinic due to **persistent stiffness and pain in the back** following a fall. Upon examination, **small swellings** around the **neck and spine** were found.

*Click to discover what
happened after finding
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Patient history

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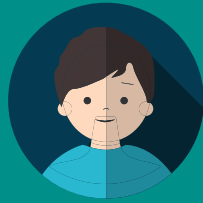
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Clinician referral

- The clinician referred the patient to a **pediatric orthopedist**

Click to discover what happened at the orthopedist





Patient history

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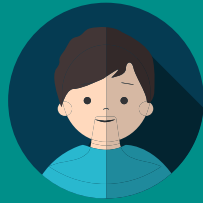
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Specialist review

- After checking the **swellings** and **medical history**, the orthopedist checked the **feet** of the patient

Click to discover what the feet revealed





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Specialist review

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Physical examination

- He found that the **two big toes** were **malformed**
- This prompted a **genetic test for FOP**, which was positive



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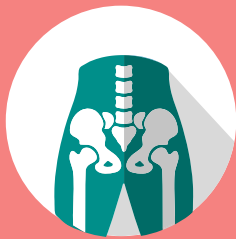
**What could
have been done
differently?**

Stretching of the jaw and muscular injury are both triggers for heterotopic ossification and should be avoided if FOP is suspected.¹



Physical features of FOP

Before heterotopic ossification becomes widespread, there are a number of physical signs that can suggest a person has FOP:¹



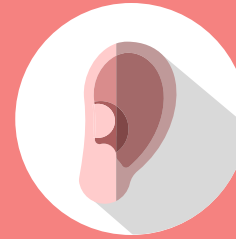
**Skeletal
malformations**

[FIND OUT MORE](#)



**Restricted
movement**

[FIND OUT MORE](#)



**Hearing
loss**

[FIND OUT MORE](#)



Flare-ups

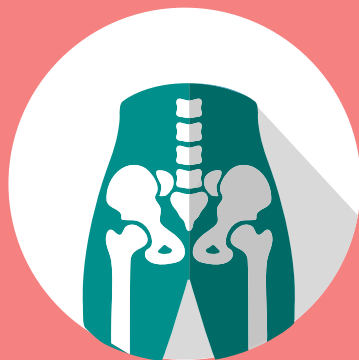
[FIND OUT MORE](#)





Physical features of FOP

Before heterotopic ossification becomes widespread, there are a number of physical signs that can suggest a person has FOP:



Skeletal malformations

- The earliest sign is malformation of the big toes, which are present in almost all classical FOP cases^{1,2}
- Similar malformation of the thumbs have been frequently observed¹
- Spinal deformation with developmental abnormalities in the cervical spine can occur¹



Physical features of FOP

Before heterotopic ossification becomes widespread, there are a number of physical signs that can suggest a person has FOP:



Restricted movement

Some babies with FOP have cervical malformations which prevent them from rotating their head completely; they may not be able to turn their head to one side. As a result, they may not be able to crawl and will scoot on their bottoms instead.^{1,2}



Physical features of FOP

Before heterotopic ossification becomes widespread, there are a number of physical signs that can suggest a person has FOP:



Hearing loss

Conductive hearing loss may occur as a result of the middle ear bones becoming restricted.^{1,2}



Physical features of FOP

Before heterotopic ossification becomes widespread, there are a number of physical signs that can suggest a person has FOP:



Flare-ups

- Flare-ups are soft-tissue swellings which are warm and painful to the touch¹
- These flare-ups often result in the ossification of surrounding structures, but in some cases they can spontaneously regress²
- Heterotopic ossification from flare-ups occurs in anatomically distinct patterns:²
 - Typically, heterotopic ossification begins in the dorsal, axial, cranial, and proximal regions²
 - As FOP progresses, heterotopic ossification may be seen in the ventral, appendicular, caudal, and distal regions²



Diagnosing FOP

ACVR1



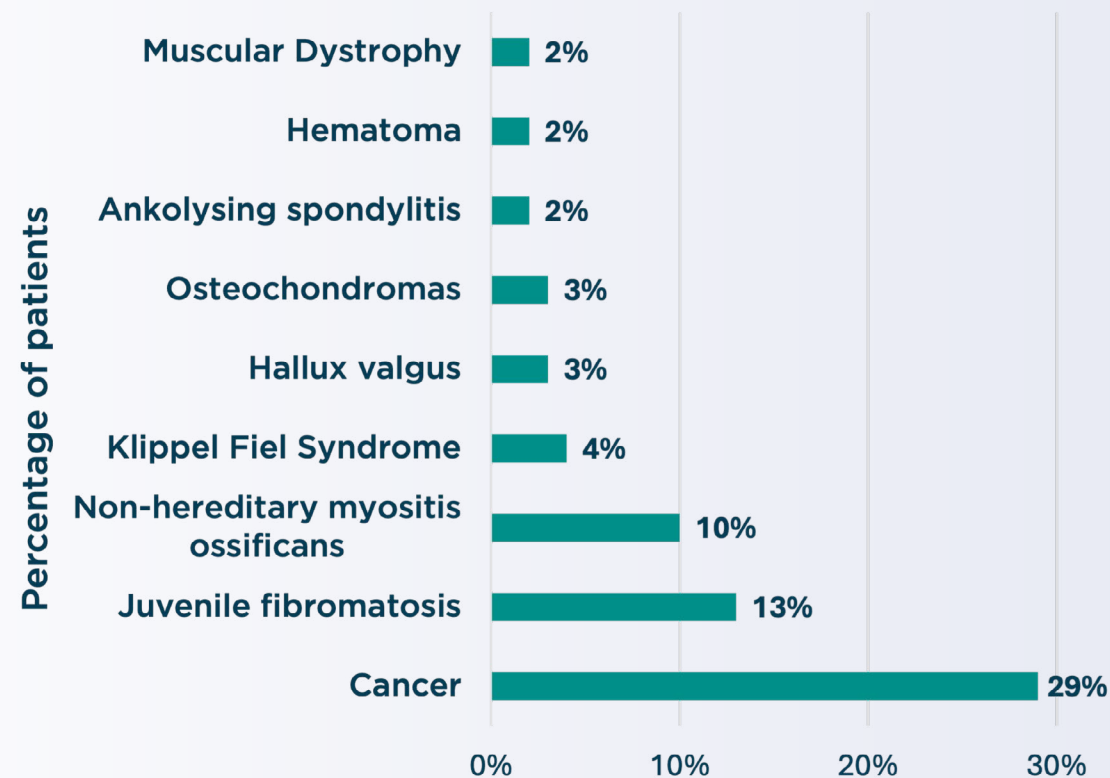
Figure adapted from Katagiri et al.¹

- FOP is caused by mutations in the ACVR1 gene, with the **617G>A** substitution being the most common²
- In cases where FOP is suspected, exon 4 of the ACVR1 gene is sequenced³
 - Changes in endonuclease restriction sites can be used as a secondary method when 617G>A is present⁴
- If 617G>A is not found, whole exome sequencing/sequencing of the full ACVR1 gene can be carried out^{5,6}



Misdiagnosis of FOP

Most Common Misdiagnoses in FOP



More than half of people with FOP have been **misdiagnosed**, with it taking an average of **1.5 years** to reach a **correct diagnosis**.¹



What are the consequences of misdiagnosis?

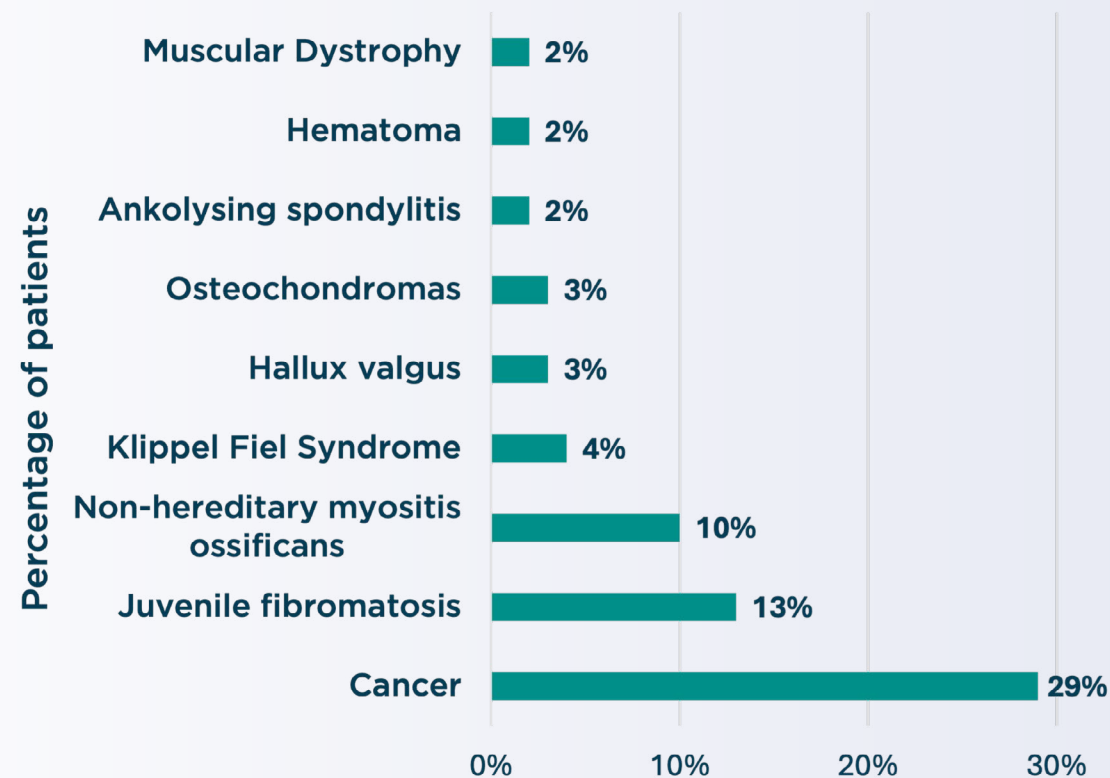
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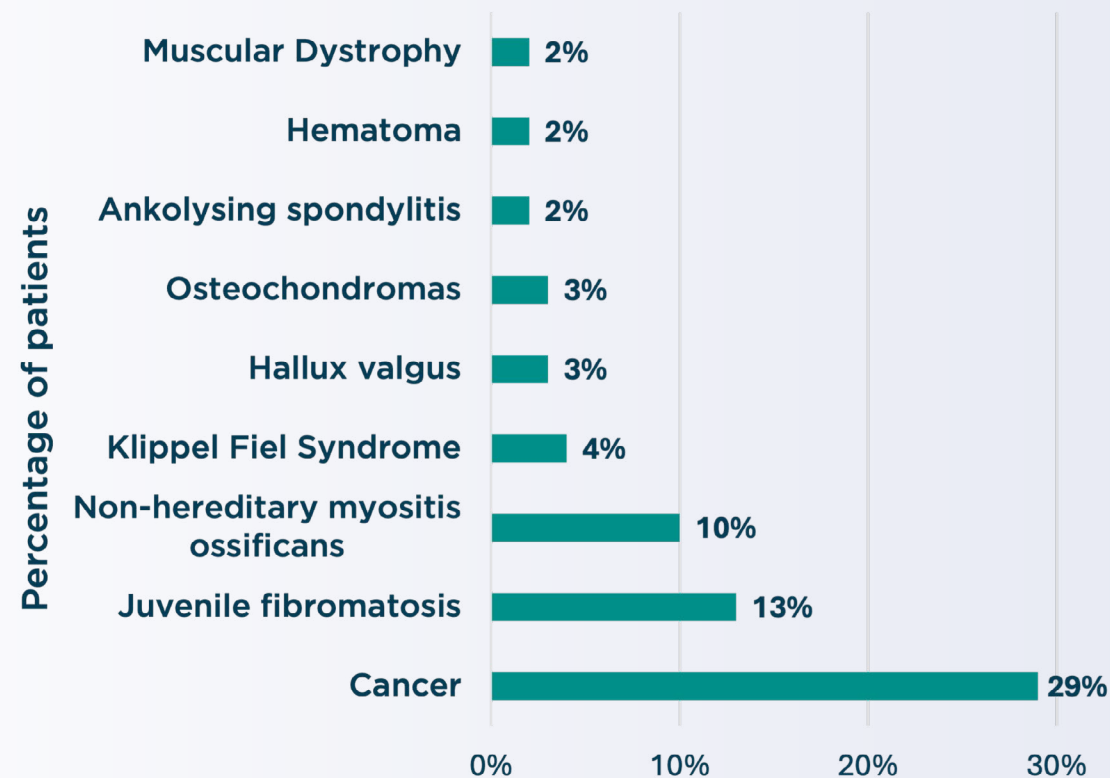
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68%
of patients receive
inappropriate therapies²



Misdiagnosis of FOP

Most Common Misdiagnoses in FOP



More than half of people with FOP have been **misdiagnosed**, with it taking an average of **1.5 years** to reach a **correct diagnosis**.¹



What are the consequences of misdiagnosis?

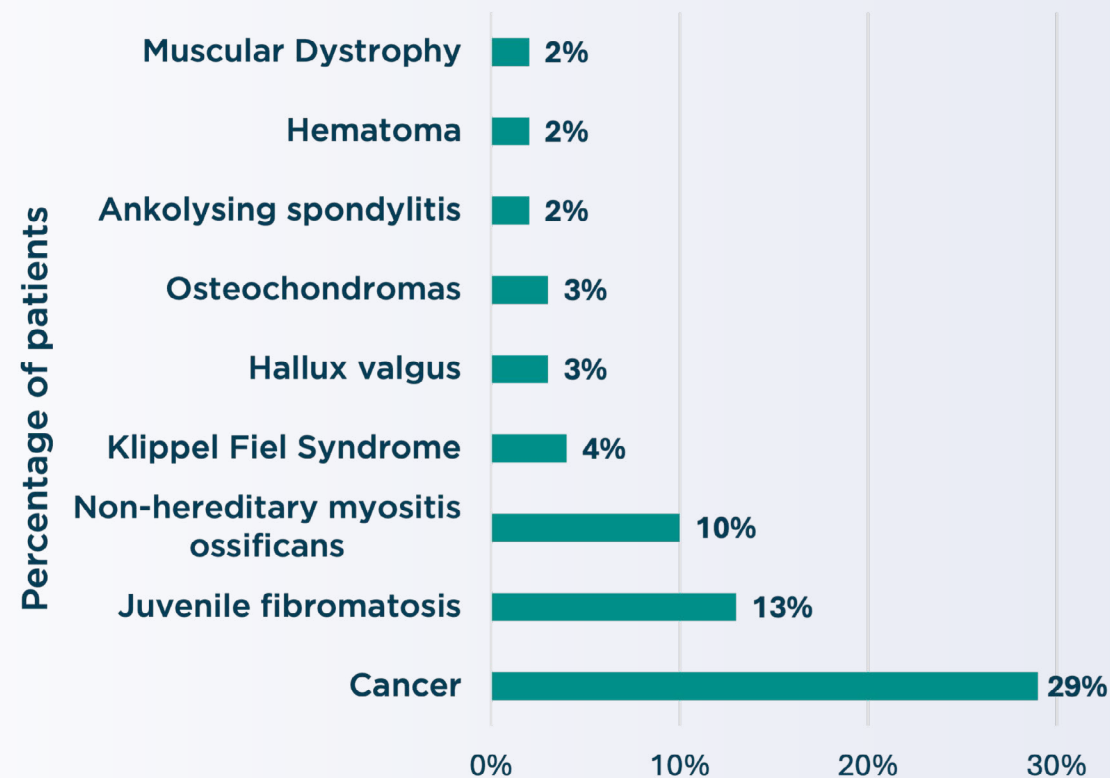
Click on the panels to discover what impact misdiagnosis of FOP can have on a patient

67%
undergo **unnecessary biopsies**²



Misdiagnosis of FOP

Most Common Misdiagnoses in FOP



More than half of people with FOP have been **misdiagnosed**, with it taking an average of **1.5 years** to reach a **correct diagnosis**.¹



What are the consequences of misdiagnosis?

Click on the panels to discover what impact misdiagnosis of FOP can have on a patient

Permanent loss of mobility following invasive interventions was reported in

49%²



Misdiagnosis of FOP

The heterotopic ossification process in FOP is progressive and cumulative; timely diagnosis can ensure harmful procedures are avoided and the patient/caregiver is educated on lifestyle changes to avoid flare-ups:

Amount of heterotopic ossification



During the **first decade**, flare-ups and **heterotopic ossification begin**.¹

During the **second decade**, joints become **ankylosed**, and many individuals **lose mobility**.²

During the **third decade**, many individuals are **wheelchair-bound** and require the aid of a caregiver.¹

During the **fourth decade**, many individuals are at risk of **early death** due to **TIS or thrombosis**.²



Preventing harm in FOP

Overexertion/
tiredness

Surgery

Dental
procedures

Biopsy

Intramuscular
injections

Muscular
stretching

Injuries

The heterotopic ossification process in FOP is progressive and cumulative; timely diagnosis can ensure harmful procedures are avoided and the patient/caregiver is educated on lifestyle changes that can be made to avoid flare-ups.^{1,2}

Click on the buttons at the top to discover more about preventing harm in FOP



Preventing harm in FOP

Overexertion/
tiredness

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Injuries

Over-exertion and fatigue can induce flare-ups, which later turn into areas of heterotopic ossification.¹



Preventing harm in FOP

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tiredness

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stretching

Injuries

Surgical release of joints is not recommended, as it risks new heterotopic ossification from the trauma.¹

If surgery is unavoidable, a multidisciplinary team with experience in managing someone with FOP is needed.²



Preventing harm in FOP

Overexertion/
tiredness

Surgery

Dental
procedures

Biopsy

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stretching

Injuries

Dental procedures, such as blocking of the nerve or stretching of the jaw muscles are not recommended, as these can cause new flare-ups to occur.^{1,2}

Preventative oral care is required in people with FOP to avoid them requiring invasive procedures later in life.²



Preventing harm in FOP

Overexertion/
tiredness

Surgery

Dental
procedures

Biopsy

Intramuscular
injections

Muscular
stretching

Injuries

Biopsies are often taken if clinicians initially suspect cancer, but this can induce a flare-up and is therefore not recommended.^{1,2}



Preventing harm in FOP

Overexertion/
tiredness

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injections

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stretching

Injuries

Intramuscular injections, including vaccines, are a risk factor for flare-ups and heterotopic ossification.^{1,2}

They should be avoided wherever possible, with alternative routes of administration (sub-cutaneous, intra-cutaneous) explored, but no immunizations should be given if the patient is experiencing a flare-up.²



Preventing harm in FOP

Overexertion/
tiredness

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Biopsy

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injections

Muscular
stretching

Injuries

Stretching of the muscles, including passive stretching to the limit of range of motion, can cause flare-ups and should therefore be avoided.¹

People with FOP should still try to maintain their physical health and function, so rehabilitation approaches such as aquatic therapy may be recommended.²



Preventing harm in FOP

Overexertion/
tiredness

Surgery

Dental
procedures

Biopsy

Intramuscular
injections

Muscular
stretching

Injuries

Muscular injury can cause flare-ups and should therefore be avoided.¹

While this means that contact sports should be avoided, people with FOP are still encouraged to exercise as much as their mobility allows, to maintain physical health (as this is often lower in people with FOP).¹⁻³